



Tangier disease

Tangier disease is an inherited disorder characterized by significantly reduced levels of high-density lipoprotein (HDL) in the blood. HDL transports cholesterol and certain fats called phospholipids from the body's tissues to the liver, where they are removed from the blood. HDL is often referred to as "good cholesterol" because high levels of this substance reduce the chances of developing heart and blood vessel (cardiovascular) disease. Because people with Tangier disease have very low levels of HDL, they have a moderately increased risk of cardiovascular disease.

Additional signs and symptoms of Tangier disease include a slightly elevated amount of fat in the blood (mild hypertriglyceridemia); disturbances in nerve function (neuropathy); and enlarged, orange-colored tonsils. Affected individuals often develop atherosclerosis, which is an accumulation of fatty deposits and scar-like tissue in the lining of the arteries. Other features of this condition may include an enlarged spleen (splenomegaly), an enlarged liver (hepatomegaly), clouding of the clear covering of the eye (corneal clouding), and type 2 diabetes.

Frequency

Tangier disease is a rare disorder with approximately 100 cases identified worldwide. More cases are likely undiagnosed. This condition is named after an island off the coast of Virginia where the first affected individuals were identified.

Genetic Changes

Mutations in the *ABCA1* gene cause Tangier disease. This gene provides instructions for making a protein that releases cholesterol and phospholipids from cells. These substances are used to make HDL, which transports them to the liver.

Mutations in the *ABCA1* gene prevent the release of cholesterol and phospholipids from cells. As a result, these substances accumulate within cells, causing certain body tissues to enlarge and the tonsils to acquire a yellowish-orange color. A buildup of cholesterol can be toxic to cells, leading to impaired cell function or cell death. In addition, the inability to transport cholesterol and phospholipids out of cells results in very low HDL levels, which increases the risk of cardiovascular disease. These combined factors cause the signs and symptoms of Tangier disease.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal

recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- A-alpha lipoprotein Neuropathy
- alpha High Density Lipoprotein Deficiency Disease
- Analphalipoproteinemia
- Cholesterol thesaurismosis
- Familial High Density Lipoprotein Deficiency Disease
- Familial Hypoalphalipoproteinemia
- HDL Lipoprotein Deficiency Disease
- Lipoprotein Deficiency Disease, HDL, Familial
- Tangier Disease Neuropathy
- Tangier Hereditary Neuropathy

Diagnosis & Management

These resources address the diagnosis or management of Tangier disease:

- Genetic Testing Registry: Tangier disease
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0039292/>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Atherosclerosis
<https://medlineplus.gov/atherosclerosis.html>
- Health Topic: Cholesterol
<https://medlineplus.gov/cholesterol.html>
- Health Topic: Diabetes
<https://medlineplus.gov/diabetes.html>
- Health Topic: Heart Diseases
<https://medlineplus.gov/heartdiseases.html>
- Health Topic: Peripheral Nerve Disorders
<https://medlineplus.gov/peripheralnervedisorders.html>
- Health Topic: Vascular Diseases
<https://medlineplus.gov/vascular diseases.html>

Genetic and Rare Diseases Information Center

- Tangier disease
<https://rarediseases.info.nih.gov/diseases/7731/tangier-disease>

Educational Resources

- Children Living with Inherited Metabolic Diseases (CLIMB): Tangier Disease
<http://www.climb.org.uk/IMD/Tango/TangierDisease.pdf>
- Disease InfoSearch: Tangier disease
<http://www.diseaseinfosearch.org/Tangier+disease/6997>
- MalaCards: tangier disease
http://www.malacards.org/card/tangier_disease
- Orphanet: Tangier disease
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=31150

Patient Support and Advocacy Resources

- American Heart Association
http://www.heart.org/HEARTORG/Conditions/Cholesterol/CholestrolATH_UCM_001089_SubHomePage.jsp
- Children Living with Inherited Metabolic Diseases (CLIMB)
<http://www.climb.org.uk/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/tangier-disease/>

Genetic Testing Registry

- Tangier disease
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0039292/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Tangier+disease%22+OR+%22Hypolipoproteinemia%22+OR+%22Tangier+Disease%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Tangier+Disease%5BMAJR%5D%29+AND+%28Tangier+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- TANGIER DISEASE
<http://omim.org/entry/205400>

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